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DATE: Wednesday, November 19, 2003

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| *DB=USPT,PGPB,EPAB,DWPI,TDBD; THES=ASSIGNEE; PLUR=YES; OP=ADJ* | | | |
| L2 | APECED and polynucleotide? and chromosome adj 21 | 4 | L2 |
| L1 | APECED same polynucleotide? and chromosome adj 21 | 2 | L1 |

END OF SEARCH HISTORY

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**Search Results** - Record(s) 1 through 4 of 4 returned.

1. Document ID: US 20010039335 A1

L2: Entry 1 of 4 File: PGPB Nov 8, 2001

PGPUB-DOCUMENT-NUMBER: 20010039335
PGPUB-FILING-TYPE: new
DOCUMENT-IDENTIFIER: US 20010039335 A1

TITLE: Secreted proteins and polynucleotides encoding them

PUBLICATION-DATE: November 8, 2001

INVENTOR-INFORMATION:

| NAME | CITY | STATE | COUNTRY | RULE-47 |
|---|---|---|---|---|
| Jacobs, Kenneth | Newton | MA | US | |
| McCoy, John M. | Reading | MA | US | |
| LaVallie, Edward R. | Harvard | MA | US | |
| Collins-Racie, Lisa A. | Acton | MA | US | |
| Evans, Cheryl | Germantown | MD | US | |
| Merberg, David | Acton | MA | US | |
| Treacy, Maurice | Co. Dublin | MA | IE | |
| Agostino, Michael J. | Andover | MA | US | |
| Steininger, Robert J. II | Cambridge | MA | US | |
| Spaulding, Vikki | Lowell | MA | US | |
| Wong, Gordon G. | Brookline | CA | US | |
| Clark, Hilary | So. San Francisco | MA | US | |
| Fechtel, Kim | Arlington | | US | |

US-CL-CURRENT: 536/23.5; 435/325, 435/69.5, 530/351

Full | Title | Citation | Front | Review | Classification | Date | Reference | Sequences | Attachments | Claims | KWIC | Draw Desc | Image

2. Document ID: US 6548272 B1

L2: Entry 2 of 4 File: USPT Apr 15, 2003

US-PAT-NO: 6548272
DOCUMENT-IDENTIFIER: US 6548272 B1

TITLE: Gene encoding for a transmembrane protein

DATE-ISSUED: April 15, 2003

INVENTOR-INFORMATION:

| NAME | CITY | STATE | ZIP CODE | COUNTRY |
|---|---|---|---|---|
| Shimizu; Nobuyoshi | Chiba | | | JP |
| Nagamine; Kentaro | Tochigi | | | JP |

US-CL-CURRENT: 435/69.1; 435/252.3, 435/254.11, 435/320.1, 435/325, 435/471, 435/71.1, 435/71.2, 530/350

Full | Title | Citation | Front | Review | Classification | Date | Reference | Sequences | Attachments | Claims | KWIC | Draw Desc | Image

3. Document ID: US 6166180 A

L2: Entry 3 of 4 File: USPT Dec 26, 2000

US-PAT-NO: 6166180
DOCUMENT-IDENTIFIER: US 6166180 A

TITLE: Chromosome 21 gene marker, compositions and methods using same

DATE-ISSUED: December 26, 2000

INVENTOR-INFORMATION:

| NAME | CITY | STATE | ZIP CODE | COUNTRY |
|---|---|---|---|---|
| Korenberg; Julie R. | Los Angeles | CA | | |
| Yamakawa; Kazuhiro | Los Angeles | CA | | |

US-CL-CURRENT: 530/350

Full | Title | Citation | Front | Review | Classification | Date | Reference | Sequences | Attachments | KWIC | Draw Desc | Image

4. Document ID: US 5773268 A

L2: Entry 4 of 4 File: USPT Jun 30, 1998

US-PAT-NO: 5773268
DOCUMENT-IDENTIFIER: US 5773268 A

TITLE: Chromosome 21 gene marker, compositions and methods using same

DATE-ISSUED: June 30, 1998

INVENTOR-INFORMATION:

| NAME | CITY | STATE | ZIP CODE | COUNTRY |
|---|---|---|---|---|
| Korenberg; Julie R. | Los Angeles | CA | | |
| Yamakawa; Kazuhiro | Los Angeles | CA | | |

US-CL-CURRENT: 435/6; 435/252.3, 435/252.33, 435/320.1, 435/325, 435/348, 435/349, 435/350, 435/352, 435/357, 435/366, 435/372.3, 514/44, 536/23.1, 536/23.5

Full | Title | Citation | Front | Review | Classification | Date | Reference | Sequences | Attachments | KWIC | Draw Desc | Image

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| Term | Documents |
|---|---|
| APECED | 78 |
| APECEDS | 0 |
| CHROMOSOME | 35012 |
| CHROMOSOMES | 18950 |
| "21" | 3009388 |
| 21S | 1119 |
| POLYNUCLEOTIDE? | 0 |
| POLYNUCLEOTIDEA | 2 |
| POLYNUCLEOTIDED | 1 |
| POLYNUCLEOTIDEJ | 1 |
| POLYNUCLEOTIDEL | 1 |
| (APECED AND POLYNUCLEOTIDE? AND CHROMOSOME ADJ 21).USPT,PGPB,EPAB,DWPI,TDBD. | 4 |

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**Search Results** - Record(s) 1 through 2 of 2 returned.

1. Document ID: US 6166180 A

L1: Entry 1 of 2 File: USPT Dec 26, 2000

US-PAT-NO: 6166180
DOCUMENT-IDENTIFIER: US 6166180 A

TITLE: Chromosome 21 gene marker, compositions and methods using same

DATE-ISSUED: December 26, 2000

INVENTOR-INFORMATION:

| NAME | CITY | STATE | ZIP CODE | COUNTRY |
|---|---|---|---|---|
| Korenberg; Julie R. | Los Angeles | CA | | |
| Yamakawa; Kazuhiro | Los Angeles | CA | | |

US-CL-CURRENT: 530/350

Full | Title | Citation | Front | Review | Classification | Date | Reference | Sequences | Attachments | Claims | KWIC | Draw Desc | Image

2. Document ID: US 5773268 A

L1: Entry 2 of 2 File: USPT Jun 30, 1998

US-PAT-NO: 5773268
DOCUMENT-IDENTIFIER: US 5773268 A

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DATE-ISSUED: June 30, 1998

INVENTOR-INFORMATION:

| NAME | CITY | STATE | ZIP CODE | COUNTRY |
|---|---|---|---|---|
| Korenberg; Julie R. | Los Angeles | CA | | |
| Yamakawa; Kazuhiro | Los Angeles | CA | | |

US-CL-CURRENT: 435/6; 435/252.3, 435/252.33, 435/320.1, 435/325, 435/348, 435/349, 435/350, 435/352, 435/357, 435/366, 435/372.3, 514/44, 536/23.1, 536/23.5

Full | Title | Citation | Front | Review | Classification | Date | Reference | Sequences | Attachments | KWIC | Draw Desc | Image

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| Term | Documents |
|---|---|
| APECED | 78 |
| APECEDS | 0 |
| CHROMOSOME | 35012 |
| CHROMOSOMES | 18950 |
| "21" | 3009388 |
| 21S | 1119 |
| POLYNUCLEOTIDE? | 0 |
| POLYNUCLEOTIDEA | 2 |
| POLYNUCLEOTIDED | 1 |
| POLYNUCLEOTIDEJ | 1 |
| POLYNUCLEOTIDEL | 1 |
| (APECED SAME POLYNUCLEOTIDE? AND CHROMOSOME ADJ 21).USPT,PGPB,EPAB,DWPI,TDBD. | 2 |

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1: Aaltonen J, Bjorses P, Sandkuijl L, Perheentupa J, Peltonen L. Related Articles, Links

An autosomal locus causing autoimmune disease: autoimmune polyglandular disease type I assigned to chromosome 21.
Nat Genet. 1994 Sep;8(1):83-7.
PMID: 7987397 [PubMed - indexed for MEDLINE]

2: Bjorses P, Aaltonen J, Vikman A, Perheentupa J, Ben-Zion G, Chiumello G, Dahl N, Heideman P, Hoorweg-Nijman JJ, Mathivon L, Mullis PE, Pohl M, Ritzen M, Romeo G, Shapiro MS, Smith CS, Solyom J, Zlotogora J, Peltonen L. Related Articles, Links

Genetic homogeneity of autoimmune polyglandular disease type I.
Am J Hum Genet. 1996 Oct;59(4):879-86.
PMID: 8808604 [PubMed - indexed for MEDLINE]

3: Aaltonen J, Horelli-Kuitunen N, Fan JB, Bjorses P, Perheentupa J, Myers R, Palotie A, Peltonen L. Related Articles, Links

High-resolution physical and transcriptional mapping of the autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy locus on chromosome 21q22.3 by FISH.
Genome Res. 1997 Aug;7(8):820-9.
PMID: 9267805 [PubMed - indexed for MEDLINE]

4: Vallada H, Craddock N, Vasques L, Curtis D, Kirov G, Lauriano V, Gentil V, Passos-Bueno R, Murray RM, Zatz M, McGuffin P, Powell JF, Gill M, Owen M, Collier DA. Related Articles, Links

Linkage studies in bipolar affective disorder with markers on chromosome 21.
J Affect Disord. 1996 Dec 16;41(3):217-21.
PMID: 8988454 [PubMed - indexed for MEDLINE]

5: Yamakawa K, Mitchell S, Hubert R, Chen XN, Colbern S, Huo YK, Gadomski C, Kim UJ, Korenberg JR. Related Articles, Links

Isolation and characterization of a candidate gene for progressive myoclonus epilepsy on 21q22.3.
Hum Mol Genet. 1995 Apr;4(4):709-16.
PMID: 7633421 [PubMed - indexed for MEDLINE]

6: Nurmi EL, Bradford Y, Chen Y, Hall J, Arnone B, Gardiner MB, Hutcheson HB, Gilbert JR, Pericak-Vance MA, Copeland-Yates SA, Michaelis RC, Wassink TH, Santangelo SL, Sheffield VC, Piven J, Folstein SE, Haines JL, Sutcliffe JS. Related Articles, Links

Linkage disequilibrium at the Angelman syndrome gene UBE3A in autism families.
Genomics. 2001 Sep;77(1-2):105-13.
PMID: 11543639 [PubMed - indexed for MEDLINE]

7: Nair RP, Stuart P, Henseler T, Jenisch S, Chia NV, Westphal E, Schork NJ, Kim J, Lim HW, Christophers E, Voorhees JJ, Elder JT. Related Articles, Links

Localization of psoriasis-susceptibility locus PSORS1 to a 60-kb interval telomeric to

HLA-C.
Am J Hum Genet. 2000 Jun;66(6):1833-44. Epub 2000 May 05. Erratum in: Am J Hum Genet 2002 Apr;70 (4):1074.
PMID: 10801386 [PubMed - indexed for MEDLINE]

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Familial sarcoidosis is linked to the major histocompatibility complex region.
Am J Respir Crit Care Med. 2000 Sep;162(3 Pt 1):861-4.
PMID: 10988096 [PubMed - indexed for MEDLINE]

9: Herr M, Dudbridge F, Zavattari P, Cucca F, Guja C, March R, Campbell RD, Barnett AH, Bain SC, Todd JA, Koeleman BP. Related Articles, Links

Evaluation of fine mapping strategies for a multifactorial disease locus: systematic linkage and association analysis of IDDM1 in the HLA region on chromosome 6p21.
Hum Mol Genet. 2000 May 22;9(9):1291-301.
PMID: 10814711 [PubMed - indexed for MEDLINE]

10: Nagamine K, Peterson P, Scott HS, Kudoh J, Minoshima S, Heino M, Krohn KJ, Lalioti MD, Mullis PE, Antonarakis SE, Kawasaki K, Asakawa S, Ito F, Shimizu N. Related Articles, Links

Positional cloning of the APECED gene.
Nat Genet. 1997 Dec;17(4):393-8.
PMID: 9398839 [PubMed - indexed for MEDLINE]

11: Oka A, Hayashi H, Tomizawa M, Okamoto K, Suyun L, Hui J, Kulski JK, Beilby J, Tamiya G, Inoko H. Related Articles, Links

Localization of a non-melanoma skin cancer susceptibility region within the major histocompatibility complex by association analysis using microsatellite markers.
Tissue Antigens. 2003 Mar;61(3):203-10.
PMID: 12694569 [PubMed - in process]

12: Yaouanq J, Perichon M, Chorney M, Pontarotti P, Le Treut A, el Kahloun A, Mauvieux V, Blayau M, Jouanolle AM, Chauvel B, et al. Related Articles, Links

Anonymous marker loci within 400 kb of HLA-A generate haplotypes in linkage disequilibrium with the hemochromatosis gene (HFE)
Am J Hum Genet. 1994 Feb;54(2):252-63.
PMID: 8304342 [PubMed - indexed for MEDLINE]

13: [No authors listed] Related Articles, Links

An autoimmune disease, APECED, caused by mutations in a novel gene featuring two PHD-type zinc-finger domains. The Finnish-German APECED Consortium. Autoimmune Polyendocrinopathy-Candidiasis-Ectodermal Dystrophy.
Nat Genet. 1997 Dec;17(4):399-403.
PMID: 9398840 [PubMed - indexed for MEDLINE]

14: Balendran N, Clough RL, Arguello JR, Barber R, Veal C, Jones AB, Rosbotham JL, Little AM, Madrigal A, Barker JN, Powis SH, Trembath RC. Related Articles, Links

Characterization of the major susceptibility region for psoriasis at chromosome 6p21.3.
J Invest Dermatol. 1999 Sep;113(3):322-8.
PMID: 10469328 [PubMed - indexed for MEDLINE]

15: Tanaka T, Ikari K, Furushima K, Okada A, Tanaka H, Furukawa K, Yoshida K, Ikeda T, Ikegawa S, Hunt SC, Takeda J, Toh S, Harata S, Nakajima T, Inoue I. Related Articles, Links

Genomewide linkage and linkage disequilibrium analyses identify COL6A1, on chromosome 21, as the locus for ossification of the posterior longitudinal ligament of the spine.

Am J Hum Genet. 2003 Oct;73(4):812-22. Epub 2003 Sep 04.
PMID: 12958705 [PubMed - in process]

16: Rubin LA, Amos CI, Wade JA, Martin JR, Bale SJ, Little AH, Gladman DD, Bonney GE, Rubenstein JD, Siminovitch KA. Related Articles, Links

Investigating the genetic basis for ankylosing spondylitis. Linkage studies with the major histocompatibility complex region.
Arthritis Rheum. 1994 Aug;37(8):1212-20.
PMID: 8053961 [PubMed - indexed for MEDLINE]

17: Scott HS, Kyriakou DS, Peterson P, Heino M, Tahtinen M, Krohn K, Chen H, Rossier C, Lalioti MD, Antonarakis SE. Related Articles, Links

Characterization of a novel gene, C21orf2, on human chromosome 21q22.3 and its exclusion as the APECED gene by mutation analysis.
Genomics. 1998 Jan 1;47(1):64-70.
PMID: 9465297 [PubMed - indexed for MEDLINE]

18: Schleutker J, Laine AP, Haataja L, Renlund M, Weissenbach J, Aula P, Peltonen L. Related Articles, Links

Linkage disequilibrium utilized to establish a refined genetic position of the Salla disease locus on 6q14-q15.
Genomics. 1995 May 20;27(2):286-92.
PMID: 7557994 [PubMed - indexed for MEDLINE]

19: Kudoh J, Nagamine K, Asakawa S, Abe I, Kawasaki K, Maeda H, Tsujimoto S, Minoshima S, Ito F, Shimizu N. Related Articles, Links

Localization of 16 exons to a 450-kb region involved in the autoimmune polyglandular disease type I (APECED) on human chromosome 21q22.3.
DNA Res. 1997 Feb 28;4(1):45-52.
PMID: 9179495 [PubMed - indexed for MEDLINE]

20: Janatipour M, Naumov Y, Ando A, Sugimura K, Okamoto N, Tsuji K, Abe K, Inoko H. Related Articles, Links

Search for MHC-associated genes in human: five new genes centromeric to HLA-DP with yet unknown functions.
Immunogenetics. 1992;35(4):272-8.
PMID: 1541487 [PubMed - indexed for MEDLINE]

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Items 1-20 of 30 Page 1 of 2 Next

1: Nat Genet. 1994 Sep;8(1):83-7.

Related Articles, Links

# An autosomal locus causing autoimmune disease: autoimmune polyglandular disease type I assigned to chromosome 21.

**Aaltonen J, Bjorses P, Sandkuijl L, Perheentupa J, Peltonen L.**

Department of Human Molecular Genetics, National Public Health Institute, Helsinki, Finland.

Autoimmune polyglandular disease type I (APECED) is an autosomal recessive autoimmune disease characterized by a variable combination of the failure of the endocrine glands. The pathogenesis of this unique autoimmune disease is unknown; unlike many other autoimmune diseases, APECED does not show association to specific HLA haplotypes. Unravelling the APECED locus will identify a novel gene outside the HLA loci influencing the outcome of autoimmune diseases. We have assigned the disease locus to chromosome 21q22.3 by linkage analyses in 14 Finnish families. Linkage disequilibrium studies have significantly increased the informativeness of the analyses and helped to locate the critical DNA region for the APECED locus to just 500 kilobases, a much more precise definition than linkage analyses alone could achieve.

PMID: 7987397 [PubMed - indexed for MEDLINE]

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**1:** Shi JD, Wang CY, Marron MP, Ruan QG, Huang YQ, Detter JC, She JX. Related Articles, Links

Chromosomal localization and complete genomic sequence of the murine autoimmune regulator gene (Aire).
Autoimmunity. 1999;31(1):47-53.
PMID: 10593569 [PubMed - indexed for MEDLINE]

**2:** Aaltonen J, Bjorses P. Related Articles, Links

Cloning of the APECED gene provides new insight into human autoimmunity.
Ann Med. 1999 Apr;31(2):111-6. Review.
PMID: 10344583 [PubMed - indexed for MEDLINE]

**3:** Mittaz L, Rossier C, Heino M, Peterson P, Krohn KJ, Gos A, Morris MA, Kudoh J, Shimizu N, Antonarakis SE, Scott HS. Related Articles, Links

Isolation and characterization of the mouse Aire gene.
Biochem Biophys Res Commun. 1999 Feb 16;255(2):483-90.
PMID: 10049735 [PubMed - indexed for MEDLINE]

**4:** Wang CY, Shi JD, Davoodi-Semiromi A, She JX. Related Articles, Links

Cloning of Aire, the mouse homologue of the autoimmune regulator (AIRE) gene responsible for autoimmune polyglandular syndrome type 1 (ASP1).
Genomics. 1999 Feb 1;55(3):322-6.
PMID: 10049587 [PubMed - indexed for MEDLINE]

**5:** Blechschmidt K, Schweiger M, Wertz K, Poulson R, Christensen HM, Rosenthal A, Lehrach H, Yaspo ML. Related Articles, Links

The mouse Aire gene: comparative genomic sequencing, gene organization, and expression.
Genome Res. 1999 Feb;9(2):158-66.
PMID: 10022980 [PubMed - indexed for MEDLINE]

**6:** Christin-Maitre S, Vasseur C, Portnoi MF, Bouchard P. Related Articles, Links

Genes and premature ovarian failure.
Mol Cell Endocrinol. 1998 Oct 25;145(1-2):75-80. Review.
PMID: 9922102 [PubMed - indexed for MEDLINE]

**7:** Bjorses P, Aaltonen J, Horelli-Kuitunen N, Yaspo ML, Peltonen L. Related Articles, Links

Gene defect behind APECED: a new clue to autoimmunity.
Hum Mol Genet. 1998;7(10):1547-53. Review.
PMID: 9735375 [PubMed - indexed for MEDLINE]

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1: Shi JD, Wang CY, Marron MP, Ruan QG, Huang YQ, Detter JC, She JX. Related Articles, Links

Chromosomal localization and complete genomic sequence of the murine autoimmune regulator gene (Aire).
Autoimmunity. 1999;31(1):47-53.
PMID: 10593569 [PubMed - indexed for MEDLINE]

2: Blechschmidt K, Schweiger M, Wertz K, Poulson R, Christensen HM, Rosenthal A, Lehrach H, Yaspo ML. Related Articles, Links

The mouse Aire gene: comparative genomic sequencing, gene organization, and expression.
Genome Res. 1999 Feb;9(2):158-66.
PMID: 10022980 [PubMed - indexed for MEDLINE]

3: Krohn K, Ovod V, Vilja P, Heino M, Scott H, Kyriakou DS, Antonarakis S, Jacobs HT, Isola J, Peterson P. Related Articles, Links

Immunochemical characterization of a novel mitochondrially located protein encoded by a nuclear gene within the DFNB8/10 critical region on 21q22.3.
Biochem Biophys Res Commun. 1997 Sep 29;238(3):806-10.
PMID: 9325172 [PubMed - indexed for MEDLINE]

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**SEARCH** Results

Documents 1 to 10 of 36563 matching the query.

**Search query:**Full Text: chromosome 21 and polyglandular Phrasing: Any

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1. **The DNA sequence of human chromosome 21**

   M. Hattori, A. Fujiyama, T. D. Taylor, H. Watanabe, T. Yada, H.-S. Park, A. Toyoda, K. Ishii, Y. Totoki, D.-K. Choi, E. Soeda, M. Ohki, T. Takagi, Y. Sakaki, S. Taudien, K. Blechschmidt, A. Polley, U. Menzel, J. Delabar, K. Kumpf, R. Lehmann, D. Patterson, K. Reichwald, A. Rump, M. Schillhabel, A. Schudy, W. Zimmermann, A. Rosenthal, J. Kudoh, K. Shibuya, K. Kawasaki, S. Asakawa, A. Shintani, T. Sasaki, K. Nagamine, S. Mitsuyama, S. E. Antonarakis, S. Minoshima, N. Shimizu, G. Nordsiek, K. Hornischer, P. Brandt, M. Scharfe, O. Schön, A. Desario, J. Reichelt, G. Kauer, H. Blöcker, J. Ramser, A. Beck, S. Klages, S. Hennig, L. Riesselmann, E. Dagand, T. Haaf, S. Wehrmeyer, K. Borzym, K. Gardiner, D. Nizetic, F. Francis, H. Lehrach, R. Reinhardt, M.-L. Yaspo
   **SUMMARY:** Chromosome 21 is the smallest human autosome. An extra copy of chromosome 21 causes Down syndrome, the most frequent genetic cause of significant mental...
   **CONTEXT:** **Chromosome 21** represents around 1-1.5% of the human genome. Since the discovery in 1959 that Down syndrome occurs when there are three copies of **chromosome 21** (ref. 1), about twenty disease loci have been mapped to its long arm, and the.....
   *Nature***405**, 311 - 319 (18 May 2000) Article
   Abstract | Full Text | PDF

2. **Autoimmune regulator: from loss of function to autoimmunity**

   J Pitkänen, P Peterson
   **SUMMARY:** The autoimmune regulator (AIRE) is a gene where mutations cause the recessively inherited disorder called autoimmune polyendocrinopathy–candidiasis–ectodermal dystrophy (APECED) or autoimmune polyendocrinopathy syndrome type...
   **CONTEXT:** ...inherited disease, autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED), also known as autoimmune **polyglandular** syndrome type 1 (APS-1).1,2 The disease usually starts with mucocutaneous candidiasis early in.....
   *Genes and Immunity***4**, 12 - 21 (13 Dec 1901) Reviews
   Abstract | Full Text | PDF

3. **Linkage disequilibrium in isolated populations: Finland and a young sub-population of Kuusamo**

   Teppo Varilo, Maris Laan, Iiris Hovatta, Victor Wiebe, Joseph D Terwilliger, Leena Peltonen
   **SUMMARY:** Linkage disequilibrium (LD), non-random association of alleles at closely linked chromosomal loci, has been used as a tool in the identification of disease alleles,...
   **CONTEXT:** markers in potential study populations can give an idea of the allelic diversity as well as of the background LD, **and** so offer some indication of the power of linkage analysis, not only for LD analysis, but also for mapping the loci.....
   *European Journal of Human Genetics***8**, 604 - 612 (01 Aug 2000) Article

   Abstract | PDF

4. **CTLA-4 in autoimmune diseases – a general susceptibility gene to autoimmunity?**

   O P Kristiansen, Z M Larsen, F Pociot
   **SUMMARY:** For most autoimmune disorders, the pattern of inheritance is very complex. The major histocompatibility complex (MHC) gene complex has been implicated as the major...
   **CONTEXT:** Genes **and** Immunity (2000) 1, 170-184 2000 Macmillan Publishers Ltd All rights reserved 1466-4879/00 $15.00 www.nature.com/gene REVIEW CTLA-4 in autoimmune diseases - a general susceptibility gene to autoimmunity? OP Kristiansen, ZM.....
   *Genes and Immunity***1**, 170 - 184 (01 Feb 2000) Review

   Abstract | PDF

5. **77 C/G mutation in the tyrosine phosphatase CD45 gene and autoimmune hepatitis: evidence for a genetic link**

   A Vogel, C P Strassburg, M P Manns
   **SUMMARY:** Autoimmune hepatitis is a chronic immune-mediated disease characterized by a loss of tolerance against liver resident antigens. The genetic background of autoimmune hepatitis is...
   **CONTEXT:** ...have been studied during the last years, including genes of human leukocyte antigens,2 cytotoxic T-lymphocyte antigen3 **and** the vitamin D receptor.4,5 In this study, we investigated an association of mutations of the CD45 molecule.....
   *Genes and Immunity***4**, 79 - 81 (13 Dec 1901) Communications
   Abstract | Full Text | PDF

6. **Familial Robertsonian translocation 15;21 and rare paracentric inv(21): unexpected re-inversion in a child with translocation trisomy 21**

   Ulrike A Mau, Uwe R Petruch, Peter Kaiser, Thomas Eggermann
   **SUMMARY:** We present a family with a Robertsonian translocation (RT) 15;21 and an inv(21)(q21.1q22.1) which was ascertained after the birth of a child with Down...
   **CONTEXT:** ...Robertsonian translocation 15;**21 and** rare paracentric inv(**21**): unexpected re-inversion in a child with translocation trisomy **21** Ulrike A Mau1, Uwe R Petruch2, Peter Kaiser1 **and** Thomas Eggermann3 P53 **and** Ras p21 proteins. In the Æve.....
   *European Journal of Human Genetics***8**, 815 - 819 (07 Nov 2000) Article

   Abstract | PDF

7. **A genome-wide scan shows significant linkage between bipolar disorder and chromosome 12q24.3 and suggestive linkage to chromosomes 1p22–21, 4p16, 6q14–22, 10q26 and 16p13.3**

H Ewald, T Flint, T A Kruse, O Mors
**SUMMARY:** The present study reports a genomewide scan using linkage analysis for risk genes involved in bipolar disorder with 613 microsatellite markers including additional testing...
**CONTEXT:** ...disorder is a severe **and** common psychiatric disorder with a lifetime risk around 0.3 1.5%. The disease is characterized by affective episodes with manic, depressive **and** other psychiatric symptoms. Mania **and** melancholia have been.....
*Molecular Psychiatry***7**, 734 - 744 (15 Aug 2002) Original Research Article
Abstract | Full Text | PDF

8. **Examination of trisomy 13, 18 and 21 foetal tissues at different gestational ages using FISH**

G E Moore, P Ruangvutilert, K Chatzimeletiou, G Bell, C-K Chen, P Johnson, J C Harper
**SUMMARY:** In man high levels of aneuploidy are seen in spontaneous abortions. Very few autosomal trisomies survive to birth, the three most common being those...
**CONTEXT:** ...of trisomy 13, 18 **and 21** foetal tissues at different gestational ages using FISH GE Moore1, P Ruangvutilert2, K Chatzimeletiou1,3, G Bell1, C-K Chen2,4, P Johnson1 **and** JC Harper2 1Department of Maternal **and** Fetal Medicine, Division.....
*European Journal of Human Genetics***8**, 223 - 228 (01 Mar 2000) Article

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9. **Paternal meiotic origin of der(21;21)(q10;q10) mosaicism [46,XX/46,XX,der(21;21)(q10;q10),+21] in a girl with mild Down syndrome**

Dieter Kotzot, Albert Schinzel
**SUMMARY:** Mosaicism for a derivative 21, der(21;21)(q10;q10), is a rare chromosomal abnormality. Since a normal cell line is present, mitotic origin is considered. Chromosome examination of...
**CONTEXT:** ARTICLE Paternal meiotic origin of der(**21**;**21**)(q10;q10) mosaicism [46,XX/46,XX,der(**21**;**21**)(q10;q10), + **21**] in a girl with mild Down syndrome Dieter Kotzot **and** Albert Schinzel Institut f¨ur Medizinische Genetik, Universit¨at Z¨urich,.....
*European Journal of Human Genetics***8**, 709 - 712 (01 Sep 2000) Article

Abstract | PDF

10. **Numerous potentially functional but non-genic conserved sequences on human chromosome 21**

Emmanouil T. Dermitzakis, Alexandre Reymond, Robert Lyle, Nathalie Scamuffa, Catherine Ucla, Samuel Deutsch, Brian J. Stevenson, Volker Flegel, Philipp Bucher, C. Victor Jongeneel, Stylianos E. Antonarakis
**SUMMARY:** The use of comparative genomics to infer genome function relies on the understanding of how different components of the genome change over evolutionary time....
**CONTEXT:** The sequence of human **chromosome 21** (ref. 8) was obtained from the National Center for Biotechnology Information (NCBI) **and** aligned with PipMaker to the mouse orthologous sequences (both sequences were hard-masked with Repeatmasker).....
*Nature***420**, 578 - 582 (05 Dec 2002) The Mouse Genome
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